ABSTRACT

The present invention relates to defects in periaxin (PRX) associated with myelinopathies, including Charcot-Marie-Tooth syndrome and/or Dejerine-Sottas syndrome. Unrelated individuals having a myelinopathy from Dejerine-Sottas syndrome have recessive *PRX* mutations. The *PRX* locus maps to a region associated with a severe autosomal recessive demyelinating neuropathy and is also syntenic to the *Prx* location on murine chromosome 7.